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TOTAL COLOBOMA OF THE OPTIC DISC WITHOUT AFFECTION OF THE CHOROID: TOTAL IRIDEREMIA: ECTROPION UVEA; AND RETAINED PUPILLARY MEMBRANE.

The Clinical Record of a Series of Congenital Malformations of the Eye.

BY

BURTON CHANCE, M. D.,

of Philadelphia.

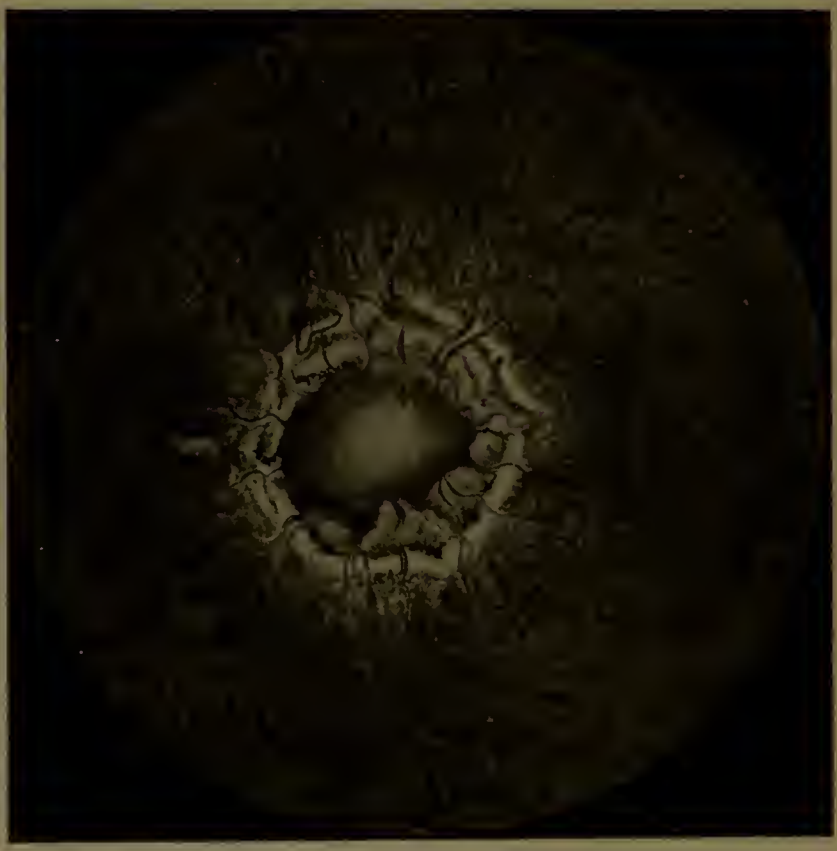
Ophthalmologist to the Germantown Hospital; Assistant Surgeon, Wills Hospital, Philadelphia.

In analyzing the records of my private practice for the past five years, it is singularly interesting to note that there have occurred several instances of congenital anomalies of the uveal tract; in these I consider myself fortunate, because in the same period I have not seen any similar cases in the public services with which I have been associated. The occurrence of anatomic congenital anomalies, as cases of unusual diseases, happens like the irony of fate; if one seeks for interesting specimens none is to be found; but when one does not expect them, they are sure to be present, and after one's neighbor has reported an instance, or at the end of a long interval in one's own experience, they come with repeated frequency. In my student days, during a ward class period, the surgeon was languishing for an opportunity to demonstrate a case of strangulated hernia. When almost in despair, a man presented himself in the waiting-room, and for six weeks thereafter the class was rewarded with strangulated hernias innumerable.

The cases of incomplete embryologic development, which are herewith recorded, are of importance, because while the specific deformities detailed are not seldom met, they illustrate conditions in single tissues unaccompanied by marked defects of contiguous structures, and are found in individuals whose personal and family histories throw no light upon the causation of deformities occurring in them.

COMPLETE COLOBOMA OF THE OPTIC NERVE WITHOUT AFFECTION OF THE CHOROID.

Localized deficiencies in the optic disc are not of infrequent occurrence. The rarest variety of coloboma of the optic nerve is that in which an enlarged and excavated disc is surrounded by a broad scleral ring, and in which the retinal vessels appear already divided in the disc and arranged regularly about its center. In



addition, as most cases of coloboma of the choroid show some involvement of the disc, so do we find associated with extensive colobomas of the disc, deficiencies, more or less complete, in the choroid and retina, in the region of the disc. Yet in 1897, Hosch,[1] of Basle, reported a case of complete coloboma of the optic disc without deformity of the choroid, and he was able to collect from

[1] Archiv für Augenheilkunde, Vol. xxiv, p. 59.

the literature only 10 other cases. The details of the case which I am about to record are in so many respects similar to that which he has reported, that they are thought to be of interest, as no other instance of this anomaly has been published since then :

A. H., aged 6, is the first child of unrelated, healthy parents, in whose families there had occurred no known case of ocular anomaly. In her early infancy it was noticed that the child's left eye turned inward, and at a later period, it became apparent that the vision of the left eye was imperfect. When first examined, the child was about 5, in the interval of several months she had learned her letters, so that the vision of the right eye is estimated at 5/10; of the left at 5/50. The right eye is hypermetropic, with +2.+0.75 ax 90=5/5; the left is myopic, with —1.25 ax 30=5/30, and accommodation most imperfect; for she reads with difficulty Snellen's type 2 D only. The child is not capable of giving intelligent replies in an attempt to outline the visual fields; the blind spot of the left eye, however, is certainly of greater dimensions than is that of the right. Color perception is manifested distinctly. The tension of each eye is normal.

The well-shaped eyes present no external abnormality. The slight internal squinting of the left eye is promptly and decidedly relieved by the use of spectacles. The hazel-gray irises are perfectly formed; under the influence of mydriatics the centrally placed pupils dilate equally and regularly.

The corneal membranes are not appreciably irregular in curvature and are free from opacities. The anterior chambers are of the usual depth. The media of both eyes are clear of opacities or extrusions. In an unfocused view, the left optic papilla appears as a great hazy mass; in the upright image the disc is about four times the diameter of the normal nerve head. It is irregularly circular in outline, and is without the usual definite rings. The arrangement here of the tissues reminds one of the appearance of a crater; or, it is like the mouth of a funnel with a broad turned-over rim. This fibrous rim is of a dense dusky whiteness, and is crimped into more or less deepened folds which are much pigmented. The central portion is indefinable, resembling a mass of cotton wool. In the inverted image it is infundibular, extending to an uncertain depth; as great an axis as —20 D can be penetrated by direct measurement.

Inside the rim of the disc is a narrow shelf-like projection of dense and darkly pigmented tissue. The sides of the funnel slope regularly, but at the apex of the cone there are no signs of the cribriform lamina. The cone is of a grayish orange color. There is neither central artery nor vein, but creeping along the sides of the cone are numerous undifferentiated vessels which are traceable until they are obscured by the overhanging border at the base of the cone. Here they are seen to wind over, bending sharply, and then to continue along the folds thereof until they finally mount over the dense ring to be distributed to the retina. There is in all, about a score of vessels, of average caliber, at the mouth of the pit, although, upon finer definition, many others of narrower caliber are observed.

The vessels disperse in much the same manner as

that depicted by Lawford,[1] and which Lindsay Johnson defines as the natural arrangement of the vessels in the eyes of bears. He also shows, in his singularly interesting atlas on the "Comparative Anatomy of the Mammalian Eye," that many other animals, chiefly the felidæ, have deep universal cups in the optic discs, and that there is an absence of the central vessels, which are replaced by a number of large vessels which run between the optic nerve and its sheath, curling over the edge of the depressed disc to spread in various directions beyond the periphery.

It is difficult to distinguish between the arteries and the veins; the arteries, however, are apparently more numerous than the veins. The veins consist of large, almost unbranched trunks extending in tortuous lines. The arteries also are unbranched, except in a few instances; they are wavy, distinctly narrower than the veins, and they taper as they approach the periphery. The temporal region of the fundus is scantily supplied with visible vessels, as quite all are found distributed to the nasal region.

The disc area resembles a yellowish crater at the summit of the reddish eyeground. The sides of this summit slope into the retinal level, the vessels bend clearly over the edges to seek the retina. Fine collections of pigment gathered together in irregular clumps form a ring encircling the nerve. A small buttonhole in the fibrous expansion on the midnasal meridian displays the underlying choroidal vessels. The macular region is of normal color, and presents no structural abnormality. In the vertical meridian, above and below the disc, the epithelium is greatly absorbed, and the choroid shines in flames like an aurora. There are no other uveal changes.

The external appearance of the right eye is normal and the media are clear. The optic disk is more pale than is usually noted in a dark-complexioned child. It is quite flush with the general retinal level, and it is vertically oval in shape, the outlines being sharply cut. It is without the usual physiologic excavation. There is a clearly defined scleral ring, beyond which is a densely black choroidal ring, the width of which varies from a quarter to a third the disc's diameter.

The entire retinal field is darkly stippled. In the vertical meridian, extending from the borders of the disc far out into the periphery, and less outlined above than below, is a pyramidal area of closely grouped collections of pigment. From the center of the disc the vessels have their entrance and exit; while they present many branchings, they are distributed only to the nasal region of the fundus, except for two tiny shoots, which go out into the temporal portion.

The nature of the disturbance which primarily affects the closure of the fetal fissure is still mysterious. As the secondary optic vessel closes the seventh week of embryonic life, it must have become active prior to that

[1] Plate 8, Vol. xv of the Transactions of the Ophthalmological Society of the United Kingdom.

period. The closure of the fissure begins at the optic nerve and ends at the ciliary body, and normally, it takes place from above, downward, and from behind, forward. Hence, the presence of colobomas of the optic nerve sheath indicates that a disturbance occurred in the vesicle at a period earlier than that which causes colobomas of the choroid. It was distinctly noticed on the examination of the present patient that there was arranged, above and below the disc, choroidal pigment in faint columns, resembling, it may be said, the markings of a raphe.

In extremely microphthalmic eyes, the nerve has been found to be imperfectly formed, and it is doubtless this imperfection has influenced the development of the eyeball. The fissure in the optic nerve stalk, may not, of necessity, close simultaneously with the closure of that in the bulb, yet, the failure to close while the choroidal fissure has disappeared remains a mystery. Can it be that there has been an antecedent inflammatory process in the intruding vitreous which has hindered the collecting of the nervous strands into a compact bundle, and has caused the sheath of the optic nerve to remain as a hollow tube, with the transmitted fibers spread out on its inner surface?

ECTROPION UVEA.

A. R., a perfectly developed, robust girl of 16, sought relief from severe headaches which were clearly dependent upon the eyestrain caused by the presence of a compound hyperopic astigmatism of two diopters. The pupils of her eyes were most irregular in outline, for extending into them and projecting into the anterior chamber were dark slaty-brown crumbly masses. These masses were not attached to the anterior surface of the iris, for during the distinctly preserved pupillary reaction, they appeared to swell, or, as de Schweinitz tersely described the condition, in his account of several similar cases which he reported before the section in Ophthalmology of the College of Physicians, a night or two before this young girl consulted me,—" they 'slopped over,' like the paint on a painter's can." In this instance the masses were larger and more numerous in the left than in the right eye. No signs of previous iritis were even present, neither was there any obstruction of the media. The choroidal ring in the right eye was very dense, being broadest along the nasal border of the disc; at the temporal side there was a moderately wide pigmented conus. The retinal epithelium was markedly granular. The left eye presented similar increase in pigmentation. The lower border of the disc was veiled by distinctly hazy fibers.

I cannot recollect any other cases of the kind since this young girl was seen, though they are sufficiently

common. Formerly, many cases of supposed neoplasms of the iris proved to be only these harmless excrescences. They are produced by an excessive and irregular growth of the uveal pigment which covers the posterior surface of the iris. Iris fibers do not enter into their structure. Having been formed, they cease to enlarge, though they sometimes, through iridic reaction, become detached and fall into the aqueous chamber where they undergo cystic degeneration.

In the horse and cow, and other allied animals, these nipple-like projections are found as proper formations, for, as these animals have large oval pupils, which contract only feebly to light, in them they serve to diminish the vertical diameter of the pupil, and thus screen the pupil from the glare.

TOTAL IRIDEREMIA.

M. E. S., aged 11. Her mother states that she had been a weakling from birth, and failed to develop as her other children. She always feared that her own depressed condition of health due to great business and domestic anxieties in the early months of her pregnancy, had profoundly impressed the unborn child, and had much to do with the child's feeble development. The birth was an easy, normal one. When about 6 this child began to have severe headaches, and it was noticed that she suffered much from the midday glare. At 7 glasses were prescribed for her by a most careful physician, but they were soon cast aside. She was a pale, thin and bony child, with a pinched face, peaked forehead, and high vertex. Although a pronounced mouth-breather, with a lofty, narrow palatal arch, there were no pharyngeal adenoids. Through her very narrow palpebral fissures, the eyes were seen to be in a state of constant and very rapid lateral nystagmus. The microphthalmic globes squinted internally, with fixation by the right.

There were sharp corneal curves. In neither eye is there any vestige of an iris. Each crystalline lens is completely formed, and is brilliantly outlined against the bright red glare from the fundus, and surrounding the lens are seen the fine radiations of the fibers of the suspensory ligament. In the right eye several fine strands like those of pupillary membranes extend from the position of the base of the iris in the lower outer quadrant, to be attached to the pole of the lens. Similar strands in the left eye were arranged in loops or festoons along the base. Pyriform opacities occupy the posterior poles of the lenses. The eyes are very hyperopic. No other embryologic or pathologic anomaly was seen in the fundus of either eye. The vision, 5/35 Sn. in each eye, is not materially improved with glasses.

The child has not been seen since the days of my examinations, for, by my advice, she was sent to school, and from 1900 she has resided at a distance. By the last accounts of her she had been keeping well up in her classes.

RETAINED PUPILLARY MEMBRANES.

C. T., a lady of advanced years, has been greatly near-sighted since childhood. In early life she suffered from an attack of granular conjunctivitis, which was followed by considerable distortion of the lids. In recent years there have been complaints of serious intraocular disturbances. The fundus of each eye shows signs of recent and remote chorioretinitis with numerous hemorrhagic foci. The margin of the right pupil is remarkably crenated, and, extending obliquely across it, from near the larger curvature of the iris, and from the upper nasal to the lower temporal quadrant, is a great broad pupillary band. There are no other congenital anomalies noticed.

I. W. H., aged 19, who resides in northern New Jersey, 12 miles from the residence of the lady whose eye has just been described, and who presented himself as I was dismissing the lady, is also a myope. In his healthy and otherwise normal right eye, are two broad pupillary bands which are arranged as the diverging sides of an angle, the apex above. The points of contact are well toward the base of the iris. The pupil is round, and reacts perfectly; during its reactions the bands become alternately taut and slack.

Just as I finished the transcription of these notes, a lady of 27 presented herself, complaining of asthenopic symptoms. In her right eye are festoons of membranous bands extending over more than the entire nasal half of the iris circle, and standing out free into the anterior chambers.